MYOT gene

myotilin

Normal Function

The *MYOT* gene provides instructions for making a protein called myotilin. Myotilin is found in heart (cardiac) muscle and muscles used for movement (skeletal muscle). Within muscle fibers, myotilin proteins are found in structures called sarcomeres, which are necessary for muscles to tense (contract). Myotilin attaches (binds) to other proteins to help form sarcomeres. Myotilin is also involved in linking neighboring sarcomeres to each another to form myofibrils, the basic unit of muscle fibers. The connection of sarcomeres to each other and the formation of myofibrils are essential for maintaining muscle fiber strength during repeated cycles of contraction and relaxation.

Health Conditions Related to Genetic Changes

limb-girdle muscular dystrophy

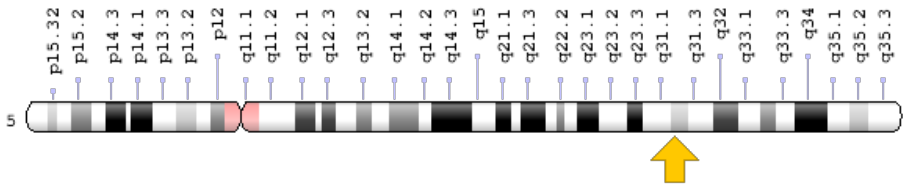
myofibrillar myopathy

At least five mutations in the *MYOT* gene have been found to cause myofibrillar myopathy. Most of these mutations are located in an area of the gene known as exon 2. *MYOT* gene mutations that cause myofibrillar myopathy change single protein building blocks (amino acids) in myotilin. Mutated myotilin proteins cluster together with other muscle proteins in the sarcomere to form clumps (aggregates). The aggregates prevent these proteins from functioning normally. A dysfunctional myotilin protein cannot properly bind with other proteins, preventing the formation of sarcomeres and myofibrils. *MYOT* gene mutations that cause myofibrillar myopathy impair the function of muscle fibers, causing weakness and the other features of this condition.

Chromosomal Location

Cytogenetic Location: 5q31.2, which is the long (q) arm of chromosome 5 at position 31.2

Molecular Location: base pairs 137,867,282 to 137,887,851 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MYOTI_HUMAN
- TTID

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Muscle Contraction https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1791
- Washington University, St. Louis: Neuromuscular Disease Center: Myofibrillar Myopathies http://neuromuscular.wustl.edu/musdist/lg.html#desmin

GeneReviews

- Limb-Girdle Muscular Dystrophy Overview https://www.ncbi.nlm.nih.gov/books/NBK1408
- Myofibrillar Myopathy https://www.ncbi.nlm.nih.gov/books/NBK1499

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MYOT%5BTIAB%5D%29+OR+%28myotilin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MYOTILIN
  http://omim.org/entry/604103

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MYOT.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MYOT%5Bgene%5D
- HGNC Gene Family: I-set domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/593
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12399
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9499
- UniProt
  http://www.uniprot.org/uniprot/Q9UBF9

Sources for This Summary

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- Schröder R, Schoser B. Myofibrillar myopathies: a clinical and myopathological guide. Brain Pathol. 2009 Jul;19(3):483-92. doi: 10.1111/j.1750-3639.2009.00289.x. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19563540
- Selcen D, Engel AG. Mutations in myotilin cause myofibrillar myopathy. Neurology. 2004 Apr 27; 62(8):1363-71. Erratum in: Neurology. 2004 Jul 27;63(2):405.
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